Desmoid tumors

A clinical review of 30 patients with more than 20 years' follow-up

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Submitted 02-07-22. Accepted 03-01-03

ABSTRACT We retrospectively studied the outcome in a consecutive series of 30 patients with desmoid tumors who were followed for more than 20 years after treatment (surgery in 29 patients). A local recurrence occurred in 12 patients and more than 1 recurrence occurred in 8. 3 patients had spontaneous complete regression of the tumor. At follow-up after mean 28 (20–54) years, all patients were tumor-free except 1 who had had a stable tumor for at least 11 years. Symptoms were related more to treatment than to the tumor. We conclude that desmoid tumors may have a high capacity for self-limitation and that conservative therapy should be considered in symptom-free patients.

In 1832, MacFarlane was the first to describe what was later named desmoid tumor or aggressive fibromatosis. It never metastasizes but may be incorrectly diagnosed as low grade fibrosarcoma. Trauma, including surgical incisions, and sex hormones have been suggested as causal factors, which may explain the reported high incidence of abdominal desmoid tumors in women after pregnancy (Stewart and Mouat 1924, Hunt et al. 1960, Dahn et al. 1963, Reitamo et al. 1986). The course of the disease varies. It may be fatal if the tumor infiltrates vital structures. Surgery has been the mainstay of treatment. Local recurrences are frequent, 19–77% (Musgrove and McDonald 1948, Hunt et al. 1960, Enzinger and Shiraki 1967, Das Gupta et al. 1968, Rock et al. 1984, Karakousis et al. 1993). Radiotherapy, chemotherapy, endocrine therapy and interferon are reported to have beneficial effects, although no conclusive evidence is available (Rock et al. 1984, Wilcken and Tattersall 1991, Kamath et al. 1996, Fernberg et al. 1999, Leithner et al. 2000). As the results of long-term treatment are rarely reported, we describe the outcome in our patients who were diagnosed more than 20 years ago.

Patients and methods

We reviewed 30 consecutive patients (20 women) with desmoid tumors seen in our Department 1949–1981 and followed for more than 20 years, mean 28 (20–54) years. 25 patients completed a standardized questionnaire, and 21 of these underwent clinical examination. In 3 patients whose clinical findings were difficult to assess MRI was done. The relatives of 2 patients who died of unrelated causes 20 and 23 years after the diagnosis, assured us that the patients had not had a recurrence. 3 patients, who had all had superficial tumors, were interviewed on the telephone and said that they were tumor-free. 2 lived far away and were unwilling to attend the outpatient clinic and 1 was too old and weak.

The tumors were located in the abdominal wall (7), upper extremity (6), lower extremity (6), back (5), thoracic wall (4), and head-and-neck region (1). 1 patient had multifocal tumors (pelvis/thigh/foot). The largest tumor diameter was mean 8 (2–16) cm.

Their mean age at diagnosis was 39 (19–67) years. All patients presented with a palpable mass. The mean duration of symptoms before the diagnosis was 13 (1– 48) months.

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Radiographic evaluation included plain radiography in 20 patients, angiography in 12, CT in 4 and MRI in 1 patient. The diagnosis was confirmed by microscopic examination before treatment in 13 patients (in 11 by fine-needle aspiration cytology and in 2 by incisional biopsy). The resected specimens were used to diagnose the other tumors. Data on the surgical margins were obtained from the surgical and/or pathological reports.

4 of the women were pregnant (3 in their first pregnancy and 1 in her sixth) or had delivered a child less than 2 years before the tumor developed. In 2 of these, the desmoid tumors were located in the abdominal wall, in 1 in the thoracic wall and 1 had multifocal tumors.

29 patients were treated surgically. 3 of these received radiotherapy later in the course of the disease because of recurrent tumors and 1 was given adjuvant antiestrogenic treatment.

One man with an abdominal tumor was treated with radiotherapy (39 Gy) alone.

Outcome (Table 1)

Continuously disease-free (CDF). 16 patients (11 female) were CDF after mean 26 (21–51) years. 1 of these patients died of a stroke 20 years after the diagnosis. 15 underwent surgical treatment. The surgical margins were wide in 9 patients and marginal in 6. 1 male patient with a desmoid tumor in the abdominal wall, which measured 10 × 8 × 7 cm, was treated with radiotherapy alone. The tumor gradually decreased in size and at follow-up 5 years later, no tumor could be seen (Figure). At the last follow-up 21 years after the diagnosis, the patient was considered to be cured. He had no symptoms after the treatment.

No evidence of disease (NED). 13 patients (8 female) had NED after mean 30 (21–54) years. 1 of these patients died of metastatic prostate cancer 23 years after the diagnosis. The surgical margin at the first operation was intralesional or marginal in all patients, except in 1 who had a wide margin. 3 patients had had 2 operations (1 a forequarter amputation). 4 patients had 3 operations (1 a lower leg amputation). 1 of these patients was also given radiation therapy after the first recurrence. 2 patients had 4 operations and 1 had 5. Radiotherapy was given after the fifth operation.

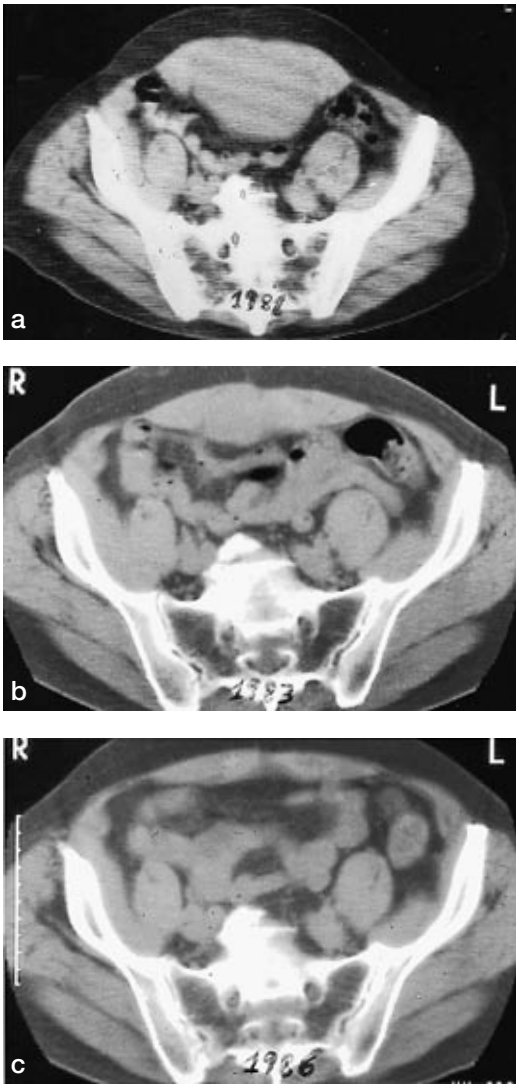
Table 1. Data in 16 CDF patients

Gender	Age	Location	Size [a]	Treatment [b]	F-U years
F	25	Abdominal wall	8	M	27
F	37	Abdominal wall	3	M	22
F	22	Abdominal wall	3	M	31
F	51	Abdominal wall	9	M	23
F	37	Back	10	M	24
F	34	Head & neck	5	M	51
F	53	Upper extremity	4	W	26
F	48	Lower extremity	6	W	30
F	54	Back	11	W	24
M	53	Back	8	W	22
M	53	Thoracic wall	14	W	22
M	60	Thoracic wall	8	W	23
F	25	Abdominal wall	2	W	23
M	35	Lower extremity	10	W	31
F	48	Lower extremity	7	W	21
M	67	Abdominal wall	10	R	21

[a] Largest diameter of the tumor (cm)
[b] M – marginal resection, W – wide resection, R – radiotherapy

The patient who first underwent a wide surgical margin because of a 7 × 5 cm desmoid tumor in the abdominal wall had a recurrence 8 months after surgery. No further treatment was given. This patient had an almost complete remission of the tumor after 5 years and at follow-up, 21 years postoperatively, the tumor had disappeared completely, which was confirmed by MRI. 2 patients were first operated on intralesionally, leaving macroscopic tumor tissue. 1 of these patients, who had a 2 cm desmoid tumor in the trapezius muscle, was operated on outside our center. The other patient had a 5 cm desmoid tumor in the shoulder region. No other treatment was given and the tumors disappeared spontaneously. At follow-up, the first patient was assessed clinically, and the other by an MRI. The follow-up data are summarized in Table 2.

Alive with disease (AWD). 1 woman is AWD. She was operated on primarily because of a 3 cm desmoid tumor in her right foot. During pregnancy 1 year later, a rapidly growing 5 cm pelvic desmoid tumor and a recurrent tumor in the right foot were found. Both tumors were resected. Postoperatively, the patient developed a recto-cutaneous fistula to the right groin, which was treated successfully with a temporary transversostomy. A recurrence of the desmoid tumor in the right groin and thigh



CT of a desmoid tumor of the rectus abdominis muscle of a 67-year-old man, who was treated with irradiation alone (39 Gy).
a. At the time of diagnosis.
b. 2 years after termination of treatment.
c. 5 years after treatment. No apparent tumor.

region occurred 3 years later. Subsequently, the patient was given radiation therapy. After 11 more years, a new solitary desmoid tumor was found in the right popliteal fossa. This tumor was operated on and local irradiation and antiestrogen therapy (Tamoxifene) were given. Two new recto-cutaneous fistulae developed in the vulva and groin region after an additional other 2 years and these were treated with a colostomy. At the last follow-up (27 years after the first surgical procedure), the desmoid tumors in the pelvis and thigh had been stable for at least 11 years, as seen on regular CT examinations. The patient had chronic recto-cutaneous fistulae, which required regular antibiotic treatment and irrigation. The range of motion in her right hip was poor. She also had lymphedema of the right lower leg and complained of persistent pain in her entire extremity.

Results of surgery. 12/27 patients developed a local recurrence. 2 still had a macroscopic tumor. The mean time until the first recurrence was 1.5 (0.3–4) years. Tumors that were excised with an intralesional or marginal margin had a greater tendency to recur (11 of 17) than those that were excised with a wide margin (1 of 10). 4 patients with desmoid tumors in the abdominal wall were operated on with a marginal margin. None of these tumors recurred.

Symptoms at follow-up. In the CDF group, 15 patients had no symptoms related to the treatment. 1 patient had slight weakness in raising of the shoulder because of removal of the accessory nerve. In the NED group, 7 patients had no symptoms, and 5 had persistent mild – moderate symptoms (reduced joint motion and/or muscle weakness). The patient who had a forequarter amputation had mild phantom pain, but did not require analgesics. The patient who was AWD had severe disabling symptoms.

No patient in our series died of their desmoid tumor.

Discussion

It is remarkable that the natural course of a tumor that was first described 170 years ago, with an annual incidence of 2–4/million in a western population, and mainly occurring in young adults, is not better understood (Reitamo et al. 1986). One reason may be that it has a very variable and unpredictable behavior. Moreover, most studies include relatively few patients with a limited follow-up and various types of treatment. The reported mortality rate is low—less than 1% (Rock et al. 1984, Karakousis et al. 1993). The ultimate fate of the patients remains unclear, which prompted us to undertake this study.

Table 2. Data in 13 patients with NED

Gender	Age	Location	Size [a]	Treatment [b]	No. of operations	F-U (years)
F	45	Back	2	I	1	36
F	29	Upper extr.	5	I	1	42
M	19	Abdominal wall	7	W	1	21
F	24	Thoracic wall	3	M/W	2	29
F	41	Upper extr.	8	M/W	2	25
M	35	Back	6	I/W	2	27
F	34	Lower extr.	5	M/W	3	26
M	52	Thoracic wall	2	M/W	3	20
F	27	Lower extr.	10	M/M + R	3	54
F	43	Upper extr.	4	I/M	3	30
M	37	Upper extr.	5	I/M	4	29
F	31	Upper extr.	5	I/W	4	28
M	28	Lower extr.	16	M/I + R	5	25

[a] Largest diameter of the tumor (cm)
[b] First operation/last operation (+ R – radiation): I – intralesional resection, M – marginal resection, W – wide resection

From the literature, it is evident that no single method of treatment is effective. The high recurrence rates after surgical excision may reflect the diffuse infiltrative growth pattern of desmoid tumors. The surgical margins may be difficult to assess, both macroscopically and microscopically. Several studies indicate that wide surgical margins are associated with a lower recurrence rate than marginal margins. However, Rock et al. (1984) reported a 50% recurrence rate following surgical excision with wide or radical margins.

In our study, only 1 of 10 patients operated on with a wide margin had a local recurrence, while 7 of 13 with marginal margins had recurrences. Altogether, our recurrence rate after primary surgery was 12/29, which is in agreement with other studies. However, it has been suggested that a recurrence could be caused by the surgical trauma itself (Reitamo et al. 1986).

The unsatisfactory results of surgical treatment and sometimes a need for disabling surgery have led to the use of other treatments. So far, radiotherapy has been reported to be the best adjuvant treatment to surgery, with reported local control rates of 79–96% (McCollough et al. 1991, Karakousis et al. 1993). However, in one report, the rate of local recurrence was 3/6 following resection and radiotherapy, as compared to 15/63 following complete excision of the tumor (Reitamo et al. 1986). Several authors have found radiation therapy to be effective not only as a complement to surgery, but also as the only treatment resulting in stable disease or tumor regression (Wara et al. 1977, Kiel and Suit 1984, Kamath et al. 1996). Nevertheless, in another report, 10 of 13 tumors in children recurred after radiotherapy (Merchant et al. 2000). Thus, the value of radiotherapy is questionable.

Antiestrogen therapy has been tried because some desmoid tumors are estrogen receptor (ER) positive (Häyry et al. 1982, Lim et al. 1986, Alman et al. 1992). In a recent study, no ER expression was found in 72 consecutive desmoid tumors, which may suggest that the sporadic effect of antiestrogen treatment on these tumors is due to other factors (Sörensen et al. 2002). In our study, only 1 patient was given additional therapy (antiestrogen), but without any obvious effect.

3 tumors disappeared spontaneously (1 recurrent tumor in the abdominal wall and 2 with a macroscopically persistent tumor). These cases resemble those reported by others (Dahn et al. 1963, Enzinger and Shiraki 1967, McDougall and McGarrity 1979, Pignatti et al. 2000), which illustrates the self-limiting characteristics of some desmoid tumors. An important question is whether this is true of all desmoid tumors. To our knowledge, no reports have been published on desmoid tumors followed without any treatment.

Most articles focus on a local recurrence, but very few report the condition of the patients at follow-up, as regards persistent symptoms or symptoms after treatment. In our study, 22 patients were symptom-free and 7 had mild–moderate symptoms. The patient with multiple tumors had severe disabling symptoms. In many patients, the symptoms may have been caused by overtreatment. The number of symptoms seemed to increase with the number of operations.

We believe that desmoid tumors with few, if any, symptoms can be followed clinically and with MRI, CT or sonography to assess their development. In stable tumors and in tumors that diminish in size, no treatment may be necessary.

No competing interests declared.

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